

## QA 40 – Galactosemia in Pregnancy

### QUESTION:

I am working with a pregnant client with galactosemia. I haven't been able to find much information on galactosemia and diet for adults. I read that up to a cup of milk each day is OK if there is some enzyme activity. However, is this contraindicated in pregnancy due to high levels of metabolites found in amniotic fluid? Any information you have to share would be appreciated.

### ANSWER:

There are several possible responses to your question:

- 1) If the woman who is pregnant has galactosemia (galactose-1-phosphate uridyl transferase deficiency, also called classic galactosemia, or GALT deficiency) it is an unusual occurrence. Because of the high frequency of ovarian atrophy (85%) most females with galactosemia do not become pregnant.<sup>3</sup>

Few pregnancies of women who have galactose-1-phosphate uridyl transferase deficiency have been reported in the literature. These women were maintained on strict lactose and galactose free diets throughout pregnancy.<sup>5</sup> It is reported that when some of these women breast-fed their infants the women developed self-intoxication with galactose probably due to lactose biosynthesis while breast-feeding.<sup>1,5</sup>

- 2) If the woman who is pregnant has the Duarte variant in which the biochemical phenotype consists of a 50% reduction in GALT enzyme activity, there is no need for dietary lactose/galactose restriction in pregnancy.
- 3) If the woman who is pregnant is a G/D (galactosemia/Duarte) compound heterozygote she would have about 25% enzyme activity and would not require dietary lactose/galactose restriction in pregnancy.
- 4) If the woman who is pregnant is a carrier for galactosemia there is no evidence that restricting lactose/galactose during pregnancy is necessary. Many people do restrict dairy products during pregnancy for 'psychological' comfort.

### References:

- 1) Brivet M, et al: Effect of Lactation in a Mother with Galactosemia. *J Pediatr* 115:280, 1989.
- 2) DeJongh S et al: Spontaneous Pregnancy in a Patient with Classical Galactosemia. *J Inherit Metab Dis* 22:754, 1999.
- 3) Kaufman F et al: Ovarian Failure in Galactosaemia. *Lancet* II: 737, 1979.
- 4) Segal S: Disorders of Galactose Metabolism. In: Scriver CR, Beaudet AI, Sly WS, Valle D: *The Metabolic and Molecular Basis of Inherited Disease*. McGraw-Hill, 1995.
- 5) Waggoner D et al: Long Term Prognosis in Galactosemia: Results of a Survey of 350 Cases. *J Inherit Metab Dis* 13: 802, 1990.